WEIGHT GAIN, DIETARY INTAKE, AND BODY COMPOSITION IN PATIENTS WITH TYPE 2 AND TYPE 3 SMA

by

Susan Peterson

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STATEMENT OF THESIS APPROVAL

The thesis of **Susan Peterson**

has been approved by the following supervisory committee members:

Kristine C. Jordan	, Chair	**04/13/2015** Date Approved
Rebecca Hurst-Davis	, Member	**04/13/2015** Date Approved
Julie Metos	, Member	**04/13/2015** Date Approved

and by **Julie Metos** , Chair/Dean of

the Department/College/School of **Nutrition**

and by David B. Kieda, Dean of The Graduate School.

ABSTRACT

Children with spinal muscular atrophy (SMA) types 2 and 3 are at risk for overweight and increased body fat. Research objectives were to determine whether increased caloric or fat intake were associated with weight increases across percentile curves over a 1 year period and to support alternative methodology for growth assessment.

This retrospective, observational study used data collected at the University of Utah. Data were reviewed one visit prior to weight increases and at subsequent visits. Dietary data from type 2 (N=16) and type 3 (N=4) participants ages 0-18 years old were analyzed. Growth status assessment included only children with type 2 SMA (N=20).

Assessments used weight-for-age percentiles, 3-day diet records and food analyses. Modified Hammersmith Functionality Scores and Compound Muscle Action Potential tracked disorder progression. Body fat percentages from dual energy x-ray absorptiometry (DXA) were compared to National Health and Nutrition Examination Survey (NHANES) percentile data and body mass index (BMI) percentiles on the Centers for Disease Control and Prevention (CDC) growth charts.

Mixed-effect analysis was used to evaluate correlations between weight increases and both diet and disorder progression. Descriptive analysis was used to assess growth chart status and body fat composition. Statistical significance was set at $p<0.05$.

There was no statistical difference in dietary intake associated with weight increases. Disorder progression was not statistically different between visits. The majority of participants were obese with body fat percentages greater than the NHANES 95th percentile at the first and last visits (12 of 20 and 19 of 20, respectively).

Results indicate that rapid weight gain in children with SMA is a product of disorder progression and not dietary alterations. Children with SMA may plot normally on the CDC BMI growth charts despite a high percent body fat. Alternative methodology for growth assessment in this population is required.

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INTRODUCTION

Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disorder resulting from a genetic mutation or deletion of the survival motor neuron (*SMN*) 1 gene on chromosome 5q13 (1, 2, 3). In a healthy population, the *SMN1* gene and several copies of the *SMN2* gene produce SMN protein, which is partially responsible for maintaining the function of motor neurons (4, 5). In persons with SMA, the mutation or deletion of *SMN1* causes significantly reduced production of SMN protein, as the *SMN2* gene can only partially compensate for the lack of *SMN1* (4). Ultimately, the homozygous mutation or deletion of *SMN1* leads to degeneration of spinal motor neurons, progressive weakness and muscular atrophy (6, 7).

The severity of SMA is generally associated with the number of *SMN2* gene copies and subsequently how much SMN protein is produced in the body. Therefore, persons with less SMN protein tend to exhibit greater severity of symptoms (2, 8, 9). Clinically, classification of the disorder is based on age of onset and the maximum functional ability achieved by the individual (10, 11). Type 1, the most severe form of SMA, is characterized by age of onset prior to 6 months of age. These individuals never develop the ability to sit unassisted. Type 2 SMA manifests between 6 and 18 months of age. These individuals are able to sit unassisted at some point during their clinical course, but are never able to walk unassisted. Type 3 SMA typically presents between 18 months

and 10 years of age. Individuals with this form of the disorder can sit, stand, and walk unassisted at some point.

Complications associated with SMA include, but are not limited to, diminished motor function, scoliosis, osteoporosis, pulmonary dysfunction, and feeding difficulties (7, 12, 13). Malnutrition, as evidenced by poor weight gain and decreased caloric intake, is most commonly seen in children with SMA type 1. This condition is of significant concern as weakened muscles may lead to dysphagia, fatigue during meals, difficulty chewing, and increased risk of choking (13, 14).

Conversely, recent studies have shown that many children with SMA types 2 and 3 may be at risk for excessive weight gain as evidenced by body fat percentage data consistent with overweight and obese (9, 15). In 2009, Sproule et al. used dual energy x-ray absorptiometry (DXA) scans to determine fat mass and fat-free mass in children (n=25) with clinically diagnosed type 1, 2, or 3 SMA (15). Results showed statistically significant reductions in fat-free mass and increases in fat mass among children with SMA compared to healthy, age-matched peers. Many subjects who had a normal body mass index (BMI) plotted between the 85^{th} and the 95^{th} percentiles for percent body fat, which was consistent with the fat-mass index percentile cutoffs "at risk for overweight" and "overweight" at the time of the study. Research by Poruk et al. in 2012 on children with SMA type 1 (n=47) showed similar results (9). Fat mass was significantly increased and fat-free mass was significantly reduced in children with SMA when compared to healthy, age-matched peers.

The research conducted by Sproule et al. and Poruk et al. indicates that there may be a number of overweight and obese patients with SMA who are undiagnosed with this

condition. Conventional methods for classifying growth patterns in a healthy population, such as the CDC growth charts, do not accurately define growth in the SMA population (16, 17). Children with SMA who are diagnosed with failure to thrive based on CDC criteria may in fact be growing normally for their age and condition; however, children with SMA who appear to be growing normally on the CDC growth chart may have levels of excess fat mass similar to overweight and obese children in a healthy population (9).

Increased fat mass consistent with overweight and obesity may negatively affect motor function in individuals with SMA (14). Muscle development is hindered in this disorder and progressive atrophy of these muscles makes them less equipped to carry excess fat. Decreased motor function and increased morbidity can result when the muscles of individuals with SMA are required to bear more weight (15). Excess fat mass is also associated with higher risk of metabolic syndrome, type II diabetes mellitus, and hypertension in children with SMA (15).

Data gathered during routine clinical visits to the Clinical Neurosciences Center show an abnormal pattern of growth in children with types 2 and 3 SMA. At a point in their development, which varies by patient, the velocity of weight gain in these children increases dramatically; weight and BMI may jump one or two percentile curves on the CDC growth charts between visits. The objectives of this research were to determine whether diet alterations, such as increased caloric or fat intake, or the progressive course of the disorder were associated with abrupt weight increases across percentile curves in these children and to provide further evidence to support alternative methodology for growth assessment in the SMA population.

METHODS

Participants

In this retrospective, observational study participants were recruited between the years 2002 and 2014 as part of the natural history study "Clinical and Genetic Studies in SMA" at the University of Utah in Salt Lake City, Utah. Age of participants ranged from birth to 18 years. Parental consent was obtained for all subjects and assent was obtained in subjects older than 7 years. This study protocol was approved by the University of Utah Institutional Review Board for Human Subjects.

Inclusion criteria for this study included a genetic confirmation of SMA 5q (*SMN1* deletion or mutation) as well as clinical diagnosis of type 2 or 3 using medically sanctioned criteria. Children with type 1 SMA were not included in this study as they have shorter lifespans and typically do not experience abrupt increases in weight percentile.

Data Collection Measures

The data obtained for this study were gathered during routine clinical visits to the Clinical Neurosciences Center. Due to the wide range of ages and types of SMA subjects, the interval between their clinical visits varied, as did the length of time they chose to continue to be treated by the SMA clinical team. Therefore, the length of participation in the study also varied among patients. Type 2 patients typically came for clinical follow-

up every 6 months, and type 3 patients' follow up visits were typically every 6-12 months. Participants were encouraged to continue with follow-up visits; however, they could discontinue their participation at any time.

Dietary Intake

To be included in this study, dietary documentation from at least two visits—one visit prior to the weight percentile increase and at least one subsequent visit until growth stabilized—had to be present. Dietary data were gathered for participants with type 2 (N=16) and type 3 (N=4) SMA. Prior to each clinical visit, parents or participants, depending on the age of the participant, were given a 3-day dietary/supplement record to complete (Appendix A). Records were to include all food, fluid, and supplements consumed during 2 weekdays and 1 weekend day. Parents and participants were given written and verbal instructions on how to complete the record (1). All diet records were analyzed for total caloric and fat intake using ESHA Food Processor (version 10.5.2, 2009 ESHA Research, Salem, Oregon).

Anthropometrics

Participants were weighed and measured during each clinical visit using standardized methods for SMA. Anthropometric measurements were completed by trained personnel. Weight was measured using an electronic scale and was documented to the nearest tenth of a kilogram. Scales used for measuring were calibrated, but the same scale was not consistently used between participants or clinical visits. Individuals who could stand on their own were measured normally on the scale, whereas individuals who

were unable to stand were measured in their wheelchairs (14). The weight of the wheelchair alone was measured and subtracted from the total weight of the participant. In order to be included in this study, participants had to exhibit an abrupt increase in weight, which was defined as an increase across one or more percentile curves on the CDC growth chart.

Height was measured segmentally with a flexible measuring tape as many participants had significant contractures (18). Segments went from head to shoulder, shoulder to hip, hip to knee, knee to ankle, and ankle to heel. Height and weight were plotted on CDC growth charts to determine any changes in growth over time.

Disorder Progression Data

Disorder progression was quantified using compound muscle action potential (CMAP) and Modified Hammersmith Functionality Scores (MHFS). CMAP is a measure of the action potential of muscle when exposed to an electrical stimulus. For the purposes of this study, CMAP amplitude, the maximum activity achieved by the muscle, and area, the total area under the action potential curve, were utilized. MHFS is a functional scoring system based on the ability to perform certain physical tasks such as the ability to sit unsupported. Disorder progression data were extracted from clinical visits for which dietary data were used.

Dual Energy X-Ray Absorptiometry (DXA)

Dual energy x-ray absorptiometry (DXA) scanning is a proven, reliable measure of changes in lean muscle mass and fat mass over time (19, 15). DXA scans were

completed using Norland DXA (XR-36 software version 3.3.1, Fort Atkinson, Wisconsin) to evaluate body composition of participants with type 2 SMA (N=20) (18). Scans were obtained at least once per year; however, researchers could ask for additional DXA scans for research purposes. Submitting to these additional scans was optional.

National Health and Nutrition Examination Survey (NHANES) Data

Body fat percentages obtained via DXA scans were categorized using smoothed percentiles for body fat percentage from a 2011 NHANES Statistics Report (20). These percentiles were compared to corresponding BMI percentiles from CDC growth charts to determine whether children with type 2 SMA fall into similar percentile curves for body fat percentage and BMI.

Demographics

Parents and participants provided demographic information as part of their consent to participate in the natural history study, "Clinical and Genetic Studies in SMA" (1). Data used in this research included age, gender, ethnicity, and race.

Statistical Methods, Data Analysis and Interpretation

Data were collected and retrieved using REDCap (Research Electronic Data Capture) (19). The Statistical Analysis Software (version 9.1.2, 2010 SAS Institute Inc, Cary, North Carolina) was used to perform all data analyses (1). Mixed-effect analysis was employed to evaluate possible correlations between abrupt increases in weight and diet and point of disorder progression. Due to the small sample size, descriptive data were

used to determine the consistency between definitions of overweight and obese based on body fat percentages from DXA scans and BMI percentiles on the CDC growth charts. The level of significance for statistical analyses was set at $p<0.05$. No power analysis was completed for this study.

RESULTS

Demographic data for all participants are presented in Table 1. Age of participants ranged from birth to 18 years, with a mean age of 4.1 years. Female participants outnumbered males by two in both the SMA type 2 and the SMA types 2 and 3 groups. The majority of participants were Caucasian.

Dietary data from 20 participants were collected for a total of 53 visits. The visits were generally 1 year apart. However, in order to increase the sample size, visits as far as 2 years apart were included if the percentile jump was substantial. Route of intake for these children included full oral feeds (n=17), partial oral feeds with supplementary formula tube feeds (n=2), and full tube feeds (n=1).

Changes in several dietary components, such as total caloric intake, total calories from fat, and fat distribution (percent calories from different fats) between visits was evaluated. Any change in diet was further compared to the increase in weight percentile to determine whether dietary modifications were correlated with weight gain. Due to large differences in caloric intake between participants with type 2 and type 3 SMA, an initial evaluation of the entire group (N=20) was compared to a second statistical analysis of participants with SMA type 2 alone (N=16). As shown in Tables 2 and 3, there was no significant change in any dietary component between visits, nor were dietary changes significantly related to an increase in weight percentile for the entire group or the type 2 group.

Disorder progression was quantified using CMAP and MHFS. Progression data were evaluated for the visit dates from which dietary data were extracted. Statistical analysis was completed to determine changes in disorder progression between visits and any change compared to the increase in weight percentile. Similar to dietary intake, there was no significant change in disorder progression measures between visits, nor were progression changes significantly related to an increase in weight percentile for the entire group or the type 2 group (see Tables 2 and 3).

Descriptive data from the comparison of patient body fat percentages obtained via DXA scans to NHANES body fat percentiles are illustrated in Figure 1. This information was further compared to the number of children with type 2 SMA (N=20) within the given BMI-for-age percentiles on the CDC growth charts. As shown in Figure 1, the majority of children had a body fat percentage falling above the NHANES 95th percentile at their first and last clinical visits (12 of 20 and 19 of 20, respectively). Conversely, relatively few children had a BMI falling above the CDC growth chart 95th percentile at their first and last clinical visits (1 of 11 for both). Participants were also more evenly distributed among the lower CDC percentiles (<50th) than the NHANES percentiles. Nine participants were missing BMI data and were not included in the CDC BMI percentile distribution analysis.

Table 1. Subject demographic data of SMA type 2 and overall (type 2 and 3)

		SMA Type 2 (N=16)	**SMA Type 2 and 3 (N=20)**
Age (yrs)	Mean (Std)	4.1 (2.0)	4.1 (2.4)
	Median (IQR)	4.1 (2.3-5.7)	3.5 (2.1-5.7)
		N (%)	**N (%)**
Gender	Male	7 (43.8)	9 (45)
	Female	9 (56.3)	11 (55)
Ethnicity	Hispanic or Latino	2 (12.5)	2 (10)
	Not Hispanic or Latino	14 (87.5)	17 (85)
	Not Reported	0 (0)	1 (5)
Race	Caucasian	15 (93.8)	18 (90)
	Asian	1 (6.3)	2 (10)

Table 2. Relationship of nutrition intake and disorder progression versus percentile increase in type 2 SMA (N=16)

		Difference between visits		Difference compared to percentile increase	
Intake/Disorder Progression Variable	Mean	Mean difference ± standard error	P-value	Mean difference ± standard error	P-value
Total caloric intake (kcal)	1078.6	27.31 ± 55.32	0.63	0.01 ± 0.01	0.37
Calories from fat (kcal)	338.9	21.49 ± 29.82	0.48	0.00 ± 0.02	0.84
Calories from saturated fat (kcal)	117.0	4.84 ± 10.70	0.66	-0.02 ± 0.04	0.72
Saturated fat distribution (% calories)	13.0	0.54 ± 1.19	0.66	-0.14 ± 0.37	0.72
Monounsaturated fat distribution (% calories)	9.5	0.51 ± 1.09	0.6	0.67 ± 0.43	0.16
Polyunsaturated fat distribution (% calories)	4.6	0.12 ± 0.59	0.85	1.10 ± 0.69	0.16
Trans fatty acid distribution (% calories)	0.4	0.19 ± 0.14	0.20	-6.55 ± 3.47	0.12
Compound Muscle Action Potential AM (mV)	2.68	0.15 ± 0.21	0.46	-0.93 ± 2.39	0.71
Compound Muscle Action Potential Area (mV)	6.13	0.11 ± 0.50	0.83	0.70 ± 1.01	0.51
Modified Hammersmith Functionality Score	19.56	-0.16 ± 0.80	0.85	0.36 ± 0.65	0.60

Table 3. Relationship of nutrition intake and disorder progression versus percentile increase in type 2 and type 3 SMA (N=20)

		Difference between visits		Difference compared to percentile increase	
Intake/Disorder Progression Variable	Mean	Mean difference ± standard error	P-value	Mean difference ± standard error	P-value
Total caloric intake (kcal)	1099.55	-3.33 ± 52.97	0.95	0.01 ± 0.01	0.32
Calories from fat (kcal)	352.15	13.78 ± 24.30	0.58	0.00 ± 0.01	0.96
Calories from saturated fat (kcal)	120.79	2.09 ± 9.00	0.82	-0.01 ± 0.03	0.84
Saturated fat distribution (% calories)	13.39	0.27 ± 1.00	0.79	-0.07 ± 0.31	0.83
Monounsaturated fat distribution (% calories)	11.02	0.58 ± 1.43	0.69	0.20 ± 0.20	0.37
Polyunsaturated fat distribution (% calories)	4.85	0.22 ± 0.55	0.69	0.98 ± 0.60	0.15
Trans fatty acid distribution (% calories)	0.49	0.11 ± 0.11	0.32	-4.87 ± 3.23	0.18
Compound Muscle Action Potential AM (mV)	3.28	0.11 ± 0.20	0.58	-1.07 ± 2.01	0.61
Compound Muscle Action Potential Area (mV)	7.64	0.22 ± 0.44	0.63	0.72 ± 0.91	0.45
Modified Hammersmith Functionality Score	21.41	-0.14 ± 0.74	0.85	0.40 ± 0.58	0.52

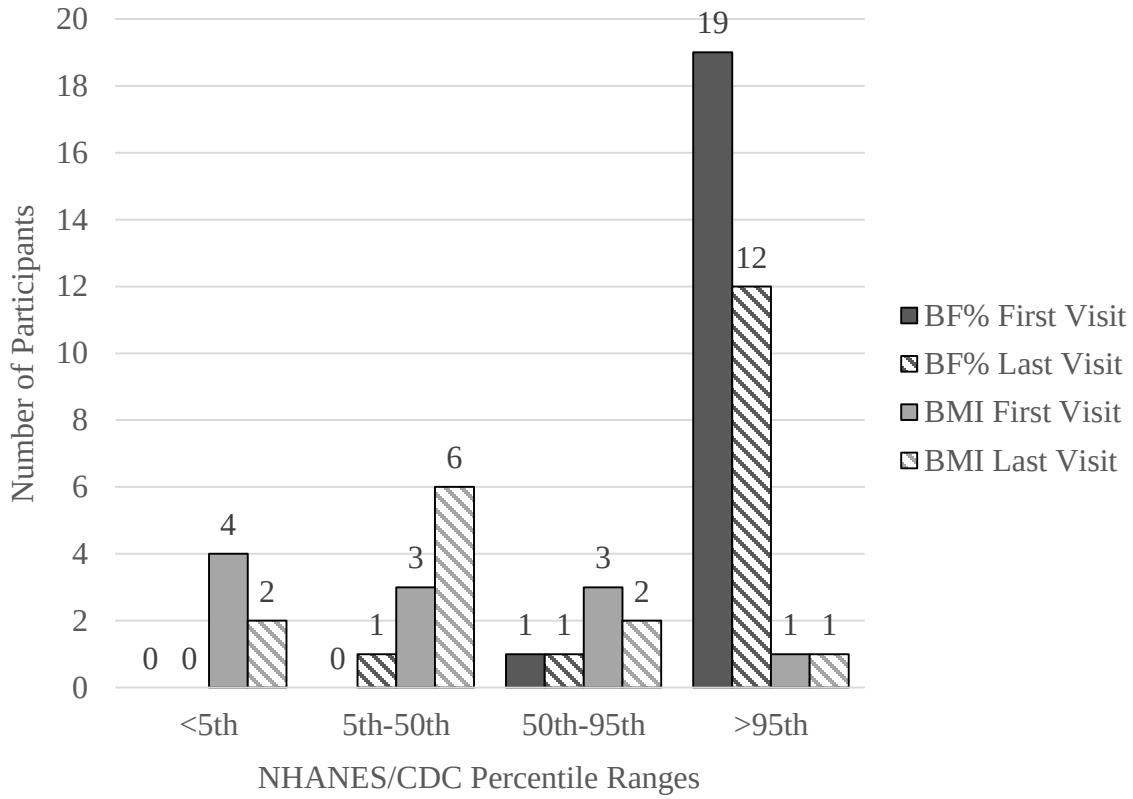

Figure 1. Distribution of participant body fat percentage (BF%) according to the NHANES percentiles compared to participant BMI according to the CDC percentiles in SMA type 2.

DISCUSSION

Children with type 2 and type 3 SMA may be at risk for overweight and obesity as evidenced by increased body fat and decreased lean body mass (15). Our study participants have demonstrated a unique growth behavior; weight may steadily follow a percentile curve for some time and then abruptly rise across percentile curves without a significant increase in caloric intake. This rapid weight increase is a cause for concern as it may speed future disorder progression and limit mobility. Furthermore, the weight gain may lead to comorbidities such as metabolic syndrome (14, 15).

This research indicates that sudden increases in weight seen in children with SMA types 2 and 3 are not likely related to alterations in caloric or fat intake. Total caloric intake did not significantly differ between visits for either the entire group or the SMA type 2 group. Similarly, calories from fat and fat distribution were not statistically different between visits for either group. Therefore, weight gain does not seem to be a product of energy input variations. We hypothesize that weight gain may rather be related to diminishing energy output over time.

Although this study did not find any significant changes in disorder progression scores between visits, SMA and disorder progression may have some association with increased weight gain. Visits were generally no more than a year apart, which may not be adequate time to detect a change in disorder progression in stable individuals with SMA.

Research by Swoboda et al. and Kaufmann et al. found that children with SMA types 2 and 3 did not show any changes in motor function after 12 months (21, 22).

Many children with SMA, especially individuals with type 2 and type 3, develop overweight and obesity according to body fat percentage (23, 24, 25). The cause of weight gain is likely multifactorial. Children with SMA have lower calorie needs (intake requirements are typically 7-14 kcal/cm height, which equates to roughly 60-80% of the RDA) than their healthy age-matched peers due to decreased energy output (25). Lean body tissue is progressively degenerated as a result of SMA. As muscle is lost, the body becomes less metabolically active and energy output decreases. Furthermore, as SMA progresses and muscle is lost, the ability to perform physical activity decreases. Both factors lead to diminishing energy output and decreasing caloric requirements over time. Thus, patients with SMA types 2 and 3 who do not alter their intake as output decreases will gain excess fat.

Recent studies with SMN-depleted mice, children with Duchenne muscular dystrophy (DMD), and children with spinal cord injuries (SCI) support this multifactorial process of weight gain. SMN-depleted mice are a useful model for the less severe forms of SMA such as type 2 and type 3 (26). Bowerman et al. found that SMN-depleted mice were generally 15-20% heavier than control mice, which researchers attributed to metabolic dyshomeostasis. DMD is a neuromuscular disorder that causes muscle weakness and atrophy similar to SMA (27). In a study from 2005, young boys with DMD had lower resting energy expenditures (REE) than their healthy age-matched peers (28). Leroy-Willig et al. also showed that higher fat mass and lower lean body mass than that of the healthy population was common in children with DMD (29). Children with SCI

develop muscular dysfunction, which causes deficits in lean muscle mass and decreased physical activity (30). A study in 2006 showed that children with SCI had significantly lower lean muscle mass and REEs that were, on average, 15.5% lower than healthy controls. Researchers hypothesized that decreased metabolic and physical activity due to muscle loss were the likely causes of body composition changes in children with DMD and SCI (27, 30).

These findings support the results of the current research. Namely, that weight gain in children with SMA is likely a result of decreased calorie needs due to lower metabolic and physical activity (disorder progression) rather than alterations in diet. Treatment of overweight and obesity requires a very delicate balance. Although excess fat mass increases disease morbidity and risk of diabetes and cardiac dysfunction, aggressive weight loss is not an effective solution. Calorie restriction may cause muscle breakdown, which is not easily regenerated (14, 15, 27). Similarly, negative nitrogen balance from inadequate protein intake can speed the process of muscular atrophy. Preventing overweight and obesity is likely the best treatment. However, treatment for children with SMA who already suffer from overweight or obesity should include an individualized plan that promotes gradual weight loss or continued growth along their established growth curve (23).

Descriptive data from Table 4 indicate that children with type 2 SMA who are categorized as overweight or obese based on body fat percentage may not be similarly categorized when using BMI and CDC growth charts. Research from children with DMD and SCI indicate that BMI is not an accurate indicator of growth status. In a study from 2003, researchers characterized 34 boys with DMD as overweight using BMI and weight

for zero muscle mass (ZMM) (31). ZMM is an equation that uses creatinine excretion to estimate lean body mass, which is then subtracted from the participant's total weight to estimate fat mass. Five participants were classified as overweight according to BMI compared to 30 participants according to ZMM, suggesting that BMI is not an accurate measurement in this population.

Research from the SCI population shows similar findings. In the study conducted by Liusuwan et al., participants with SCI had a similar BMI to the healthy controls despite having a significantly higher percent body fat (30). BMI is not a sensitive indicator of body composition and growth, as it does not distinguish between fat mass and fat-free mass. Researchers have seen statistically significant reductions in fat-free mass and increases in fat mass among children with SMA type 2 and 3 compared to healthy, age-matched peers (15). As indicated by the current study and previous research, BMI is not an effective measurement of growth status and cannot be used to diagnose overweight and obesity in children with SMA.

Limitations

A limitation of this study is the small sample size. Many children with SMA types 2 and 3 showed the pattern of weight gain described in this study but lacked dietary or disorder progression data for the visit dates in which the weight gain was seen. Additionally, some study candidates showed an increase in weight but it manifested between visit dates that were too many years apart. The conclusions of this research are limited due to the small number of participants.

Potential error in dietary documentation and anthropometric measurements is a second limitation of this study. Three-day diet records were completed by parents or the participants rather than a dietitian or other member of the SMA clinical team. Parents and participants may document intake with a low level of specificity, which makes accurate dietary analysis difficult. Similarly, there was potential for error in anthropometric measurements. In particular, accurate measurements of height may be difficult to obtain if the child has contractures. The scales used to measure weight varied between patient and clinical visit, which possibly decreased the accuracy of this measure.

The use of descriptive statistics when comparing NHANES body fat percentiles to BMI percentiles on the CDC growth chart is another limitation. Although this information is useful, it lacks statistical power. The NHANES Statistical Report from 2011 also used a GE Holistic DXA machine, whereas researchers in this study used a Norland DXA machine. The use of different machines may cause discrepancies between the body fat percentages used to create the NHANES percentiles and the body fat percentages obtained for the participants of this study.

Conclusion

Our study results support current knowledge of overweight and obesity in children with SMA and neuromuscular conditions. The development of excess fat mass in children with SMA type 2 and type 3 is most likely secondary to the progressive nature of SMA rather than alterations in caloric or fat intake. Research to determine the causes of these sudden increases in weight across percentile curves is necessary. This information could improve clinical care for patients by suggesting methods for

preventing the development of overweight and obesity. Also, established guidelines for nutrition management in people with SMA who already suffer from excess fat are essential as aggressive weight-loss measures can result in further muscle loss. Small, incremental decreases in calories may be beneficial. Finally, this study adds support for the establishment of alternative methodology for growth assessment in the SMA population.

APPENDIX: FOOD RECORD

FOOD RECORD

Child's name: _____

Name of the caregiver(s) completing this form:

_____

Relationship to the child:

_____

Date:

_____

Phone contact information if needed for clarification:
___Cell:_____
___Home:_____
___Work:_____

My child's feeds are:
___All by mouth
___By mouth and by feeding tube
___Restricted to feeding tube only

Please return this form to:

Dr. Kathryn Swoboda
Pediatric Motor Disorders Research Program
Department of Neurology
University of Utah School of Medicine:
50 North Medical Drive, Room 3R210
Salt Lake City, Utah 84132
FAX: 801-587-9346

Please contact any of our clinical coordinators for questions regarding completion of this form:
801-585-9717

Nutritional Care Guidelines for children with SMA and other neuromuscular disorders are available on our web site: http://medicine.utah.edu/neurology/research/swoboda

DIRECTIONS FOR KEEPING THIS FOOD DIARY:

Please record everything your child eats or drinks over a three day period, preferably two weekdays and one weekend day. This record should include feeds and supplements and all water or other liquids given via mouth, nasogastric, nasojejunal or gastrostomy tube. If your child consumes the identical diet each day via tube-feeding, just indicate this, and completion of only a single day is necessary.

1. Record all food and drink whether eaten/given at home or away from home.

2. Be as specific as possible in recording the item, and use a brand name if available and include a label for nutrient information.

3. Measure the item when possible or estimate the amount using the pictures provided with this document. You can record in cubic centimeters (cc) or ounces (oz) with regard to liquids, and either ounces (oz) or grams (g) for food items.

4. Please mention if possible how the food was prepared (fried, boiled, baked, microwaved, etc) and record items used in the preparation, including oil, margarine or butter, for instance.

5. Don't forget to include all condiments, such as catsup, gravy, sauce, butter or oil, and record amounts as accurately as possible. For instance, you might record 2 french fries with 1 teaspoon catsup.

6. Please include one item per line where possible.

7. Please include all supplements (vitamins) given, with brand names and labels if possible.

8. You can use abbreviations for measurements:
Teaspoon – tsp ¼ cup – ¼ C
Tablespoon – Tbsp Ounce – oz

9. For infants, record the duration and frequency of breast feeds, as well as the pre- and post-feeding weight of your infant one time per day.

EXAMPLE: INFANT'S DIET
Try to record only what is eaten (and not what falls in the lap or on the floor)

Dinner: 1 x 4 oz jar stage I pears (Gerber)
6 ounces Carnation Goodstart Formula

EXAMPLE: INFANT'S DIET, GASTROSTOMY TUBE WITH BOLUS FEEDING

Lunch 1 x 4 oz jar stage I peaches (1 Tbsp by mouth, the rest via G-tube)
6 oz formula (Pediatric Vivonex Standard dilution for 20 calories/oz)

Overnight continuous feeds with pediatric Vivonex at 30 cc/hour x 8 hours

EXAMPLE: INFANT'S DIET: GASTROSTOMY TUBE ONLY, CONTINUOUS FEEDS

Continuous feeds: 45 cc/hour, pediatric Vivonex standard dilution
OR
Continuous feeds: 45 cc/hour, Tolerex formula with added nutrients including the following.
. .
Total recipe = (please include all ingredients including added vitamins, oil, juices and any supplements)

EXAMPLE: TODDLER'S DIET
Try to record only what is actually eaten

Dinner: 2 Tbsp stage II squash (Gerber)
½ C whole milk
1 oz chicken (2 pieces of chicken tenders, brand name, baked)
5 small pieces of cantaloupe (grape sized)
¼ cup macaroni and cheese (Annie's brand, prepared as directed)

PLEASE CIRCLE DAYS RECORDED AND DATES

SUN MON TUES WED THURS FRI SAT
DATE(S):_____

FOR SPECIAL FORMULA MIXTURES, PLEASE LIST ALL INGREDIENTS HERE:

FOR ADDITIONAL VITAMIN, MINERAL OR HERBAL SUPPLEMENTS OF ANY TYPE, INCLUDING PROTEIN OR AMINO ACID SUPPLEMENTS: please list brand name if possible, full name and attach label if possible:

BREASTFEEDING: please list approximate time and duration of feeding in the table below. If you think you can estimate how much your baby receives in terms of ounces or cc's, please do so. If you pump breastmilk, this can provide some estimate of how much your baby might be getting per feed. If your baby feeds frequently for short periods of time, estimate total amount of feeding time in a 24 hour period if you can.

DAY 1: DATE/TIME	**FOOD, FORMULA OR FLUID**, including details of preparation if not provided above, or for powdered formulas, note calories/oz prepared	**AMOUNT OR RATE (IF FEEDING PUMP USED)**

For breastfed infants, choose one feed per day: **Pre-feeding weight: _____lbs or kg**
Post feeding weight: _____lbs or kg
Duration of feeding: _____

DAY 1: DATE/TIME	**FOOD, FORMULA OR FLUID**, including details of preparation if not provided above, or for powdered formulas, note calories/oz prepared	**AMOUNT OR RATE (IF FEEDING PUMP USED)**

For breastfed infants, choose one feed per day: **Pre-feeding weight: _____lbs or kg**
Post feeding weight: _____lbs or kg
Duration of feeding: _____

DAY 1: DATE/TIME	**FOOD, FORMULA OR FLUID**, including details of preparation if not provided above, or for powdered formulas, note calories/oz prepared	**AMOUNT OR RATE (IF FEEDING PUMP USED)**

For breastfed infants, choose one feed per day: **Pre-feeding weight: _____lbs or kg**
Post feeding weight: _____lbs or kg
Duration of feeding: _____

REFERENCES

1. Aton J, Davis RH, Jordan KC, et al. Vitamin D intake is inadequate in spinal muscular atrophy type I cohort: correlations with bone health. Journal of Child Neurology 2013;0:1-7.

2. LeFebvre S, Burgien L, Reboullet S, et al. Identification and characterization of spinal muscular atrophy-determining gene. Cell 1995;80:155-65.

3. Wirth B, Herz M, Wetter A, et al. Quantitative analysis of survival motor neuron copies: identification of subtle *SMN 1* mutations in patients with spinal muscular atrophy, genotype-phenotype correlation, and implications for genetic counseling. American Journal of Human Genetics 1999;64:1340-56.

4. Lunn MR, Wang CH. Spinal muscular atrophy. Lancet 2008;371:2120–33.

5. Markowitz JA, Singh P, Darras BT. Spinal muscular atrophy: a clinical and research update. Pediatric Neurology 2012;46:1-12.

6. Oskoui M, Levy G, Garland CJ. The changing natural history of spinal muscular atrophy type 1. Neurology 2007;69:1931-6.

7. Sproule DM, Montes J, Dunaway S, et al. Adiposity is increased among high-functioning, non-ambulatory patients with spinal muscular atrophy. Neuromuscular Disorders 2010;20:448–52.

8. Feldkotter M, Schwarzer V, Wirth R, et al. Quantitative analyses of *SMN1* and *SMN2* based on real-time lightCycler PCR: fast and highly reliable carrier testingand prediction of severity of spinal muscular atrophy. American Journal of Human Genetics 2002;70:358-68.

9. Poruk KE, Davis RH, Smart AL. Observational study of caloric and nutrient intake, bone density, and body composition in infants and children with spinal muscular atrophy type I. Neuromuscular Disorders 2012;22:966–73.

10. Munsat TL. Workshop report, International SMA collaboration. Neuromuscular Disorders 1991;1:81.

11. Tsirikos AI, Baker ADL. Spinal muscular atrophy: classification, aetiology, and treatment of spinal deformity in children and adolescents. Current Orthopaedics 2006;20:430–45.

12. Mercuri E, Bertini E, Iannaccone ST. Childhood spinal muscular atrophy: controversies and challenges. Lancet Neurology 2012;11:443–52.

13. Messina S, Pane M, De Rose P, et al. Feeding problems and malnutrition in spinal muscular atrophy type II. Neuromuscular Disorders 2008;18:389-93.

14. Sproule DM, Hasnain R, Koenigsberger D, et al. Age at disease onset predicts likelihood and rapidity of growth failure among infants and young children with spinal muscular atrophy types 1 and 2. Journal of Child Neurology 2012;27:845-51.

15. Sproule D, Montes J, Montgomery M, et al. Increased fat mass and high incidence of overweight despite low body mass index in patients with spinal muscular atrophy. Neuromuscular Disorders 2009;19:391–6.

16. Frisancho AR. Anthropometric standards for the assessment of growth and nutritional status. Ann Arbor, MI, University of Michigan Press 1993.

17. Tilton AH, Miller MD, Khoshoo V. Nutrition and swallowing in pediatric neuromuscular patients. Seminar in Pediatric Neurology 1998;5:106-15.

18. Hurst Davis R, Zhang Y, Swoboda KJ. Arm anthropometry is associated with body composition in patients with spinal muscular atrophy. Currently under review for publication.

19. Chan G. Performance of dual-energy x-ray absorptiometry in evaluating bone, lean body mass, and fat in pediatric subjects. Journal of Bone and Mineral Research 1992;7:369-74.

20. Ogden CL, Li Y, Freedman DS, et al. Smoothed percentage body fat percentiles for U.S. children and adolescents, 1999-2004. National Health Statistics Report 2011;43:6-7.

21. Swoboda KJ, Prior TW, Scott CB, et al. Natural history of denervation in SMA: relation to age, SMN2 copy number, and function. Ann Neurol 2005;57:704-12.

22. Kaufmann P, McDermott MP, Darras BT, et al. Observational study of spinal muscular atrophy type 2 and 3: functional outcomes over 1 year. Arch Neurol 2011;68:779-86.

23. Wang CH, Finkel RS, Bertini ES, et al. Consensus statement for standard of care in spinal muscular atrophy. Journal of Child Neurology 2007;22:1038-41.

24. DiVito D, Konek S. Spinal muscular atrophy: summary for nutritional care. Infant, Child, and Adolescent Nutrition 2010;6:348-54.

25. Godshall B, Wong B. Spinal muscular atrophy: an overview of disease and nutrition. PNPG Building Block 2012;1:1-4.

26. Bowerman M, Michalski J-P, Beauvais A, et al. Defects in pancreatic development and glucose metabolism in SMN-depleted mice independent of canonical spinal muscular atrophy neuromuscular pathology. Human Molecular Genetics 2014;23:3432-44.

27. Davidson ZE, Truby H. A review of nutrition in Duchenne muscular dystrophy. Journal of Human Nutrition and Dietetics 2009;22:383-93.

28. Gonzalez-Bermejo J, Lofaso F, Falaize L, et al. Resting energy expenditure in Duchenne patients using home mechanical ventilation. European Respiratory Journal 2005;25:682-7.

29. Leroy-Willig A, Willig TN, Henry-Feugeas MC, et al. Body composition determined with MR in patients with Duchenne muscular dystrophy, spinal muscular atrophy, and normal subjects. Magnetic Resonance Imaging 1997;15:737-44.

30. Liusuwan RA, Widman LM, Abresch RT, et al. Body composition and resting energy expenditure in patients aged 11 to 21 years with spinal cord dysfunction compared to controls: comparisons and relationships among the groups. Journal of Spinal Cord Medicine 2007;30:S105-11.

31. Pessolano FA, Suarez AA, Monteiro SG, et al. Nutritional assessment of patients with neuromuscular diseases. American Journal of Physical Medicine and Rehabilitation 2003;82:182-5